**MODULE 27 - ETIOLOGIES**

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INTRODUCTION

Mental retardation is a term used in today’s society that describes a group of people who are unable to function effectively due to intellectual and adaptive behavioral deficiencies. Historically, this group has been ignored, exploited, and often scorned as fools or simpletons.

Jean Itard undertook the first systematic study of a mentally retarded person. Itard’s work on the “Wild Boy of Aveyron” is a classic study on the education of children deprived of early human stimulation. Other clinicians and educators undertook to explore new methods for improving the quality of life for these individuals. Edouard Sequin, a former student of Itard, in the 1830’s and 1840’s, developed a variety of systematic procedures for retraining the basic perceptual and motor functions of the retarded. During this same period, several institutions were opened, devoted to the custodial care and treatment of retarded people. By 1876, there were 12 such residential schools in the United States.

Advances were slow in the next seventy-five years. Few programs were involved in the prevention of mental retardation or rehabilitation of clients. Most of the care remained custodial. It has been since 1950 that major changes and advances have occurred in the treatment and training of the developmentally disabled individual.

A useful guide in research, prevention, and the treatment of mental retardation is a system of classification, developed by the American association on Mental Retardation -(AAMR). There is still confusion, however, in differentiating and grouping the many varieties of mental retardation. This module focuses on “etiologies and classifications” of the developmentally disabled. Also included is content on epilepsy, cerebral palsy, and genetic counseling. However, the candidate is reminded that mental retardation does not necessarily accompany epilepsy or cerebral palsy.
OBJECTIVES

THEORY: The successful candidate will achieve a passing score (75%) on a written comprehensive examination covering the etiologies and classifications of developmental disabilities according to the American Association of Mental Retardation (AAMR).

ASSESSMENT: There will be a written comprehensive test: multiple choice, true/false, and matching questions.

MAKE UP TESTS MAY BE AN ESSAY TEST!!

INSTRUCTIONAL MEDIA: Study Guides

1. Purposes of assessment
2. Components of assessment
3. Examples of assessment
4. Factors in intervention

INSTRUCTIONAL MEDIA:


Textbook:

Beirne-Smith

1. Chapter 6: Biological Aspects and the Promises of Prevention

Study guide 1: Definition and classification of Mental Retardation.

The successful candidate will be able to:

1. Identify the four (4) gradations of developmental disabilities as defined by the AAMR.
2. Identify the incidence or occurrence of developmental disabilities in the United States.
3. Match the estimated level of retardation with the expected level of adaptive behavior for a given age group.
4. Match intelligence quotients to levels of adaptive behavior.
5. Identify the estimated level of retardation for each of the following educational terms:
   a. Educable
   b. Trainable
   c. Dependent (Custodial)

INSTRUCTIONAL MEDIA:

Study Guide 2: Etiologies

The successful candidate will be able to:

1. Identify the ten (10) major medical classifications of mental retardation based on the etiological groups as defined by the AAMR.
   a. Infections and Intoxications
   b. Trauma or Physical Agents
   c. Metabolism or Nutrition
   d. Gross Brain Disease
   e. Unknown Prenatal Influences
   f. Chromosomal Abnormalities
   g. Gestational Disorders
   h. Psychiatric Disorders
   i. Environmental Disorders
   j. Other Conditions

INSTRUCTIONAL MEDIA:

Study Guide 2: Etiologies

The successful candidate will be able to:

1. Using the AAMR classification of retardation, match the major classifications with their specific disorders.
   a. Prenatal Intoxications
   b. Trauma or Physical Agents
   c. Metabolism or Nutrition
   d. Gross Brain Disease
   e. Unknown Prenatal Influences
   f. Chromosomal Abnormalities
   g. Gestational Disorders
   h. Psychiatric Disorders
2. Identify the major diagnostic features, descriptions, and/or treatment for each of the following conditions.

   a. Autism
   b. Battered-Child Syndrome
   c. Cretinism
   d. Down’s Syndrome
   e. Hurler’s Syndrome
   f. Hydrocephalus
   g. Kernicterus
   h. Myelomeningocele
   i. Phenylketonuria
   j. Rubella
   k. Sturge-Weber
   l. Tay-Sachs
   m. Tuberous Sclerosis

INSTRUCTIONAL MEDIA:

Study Guide 3: Cerebral Palsy

The successful candidate will be able to:

1. Identify the accepted definition for cerebral palsy.
2. Identify the five (5) types of cerebral palsy with their appropriate definitions.
3. Match each of the five types of cerebral palsy with their appropriate descriptions.
4. Identify three prenatal causes of cerebral palsy.
5. Identify four causes of cerebral palsy that occur during the developmental period.
6. Identify associated handicaps that often occur with cerebral palsy.
7. Identify treatments used for the movement disorders of cerebral palsy.
8. Identify tests used to diagnose cerebral palsy, with appropriate descriptions of each.
INSTRUCTIONAL MEDIA:

Study Guide 4: Epilepsy

The successful candidate will be able to:

1. Define epilepsy according to your module.

2. Match each of the major types of seizures with their appropriate definitions.
   a. Grand mal seizures
   b. Petit mal seizures
   c. Psychomotor seizures
   d. Focal motor seizures
   e. Focal sensory seizures

3. Identify the appropriate definition for an “aura”.

4. Identify the methods used in the diagnosis of epilepsy.

5. Identify conditions, which can increase the frequency of seizures.

INSTRUCTIONAL MEDIA:

Study Guide 5: Genetic Counseling

The successful candidate will be able to:

1. Identify the accepted definition of genetic counseling.

2. Identify basic steps used in the process of counseling parents:
   a. Establishing an accurate diagnosis.
   b. Securing a detailed family history.
   c. Providing up-to-date genetic information.
   d. Establishing rapport with the client.

3. Identify the most common diagnostic test/method used in genetic counseling.
PRINCIPLES

1. A classification is a systematic arrangement of books, documents, archives, and other printed material in accordance with categories.

2. Etiology is the giving of a cause or reason for anything. It can also be the reason itself.

3. An individual who is significantly sub average in general intellectual functioning with deficits in adaptive behavior manifested in the developmental period is classified as mentally retarded.

4. Classification is an important tool used in clinical analysis and is a useful guide in research, prevention and treatment of the mentally retarded.
VOCABULARY

Adaptive Behavior
Amniocentesis
Ataxia
Athetosis
Autosomal dominant
Buphthalmos
Café-au-lait-spots
Choreo-athetosis
Chorioretinitis
Chromosomes
Concomitant
Custodial
Developmental Period
Disease
Educable
Encephalophathy
Epicanthal Folds
Etiologies
Genes
Genetic
Hepatomegaly
Hirsutism
Hypertelorism
Inheritance
Intellectual Functioning
Kernicterus
Karyotype
Mental retardation
Metabolism
Muscle dysfunctions
VOCABULARY

Osteochondritis
Psychomotor
Rigidity
Sensory-Motor
Spasticity
Symbiotic
Syndrome
Trainable
Tremor
Trisomy
Venous Angioma
STUDY GUIDE I
DEFINITION AND CLASSIFICATION OF MENTAL RETARDATION

As defined by the American Association on Mental Retardation (AAMR) mental retardation refers to significantly sub average general intellectual functioning existing concurrently with deficits in adaptive behavior, and maturational development, and manifested during the developmental period.

GENERAL INTELLECTUAL FUNCTIONING is defined as the results obtained by assessment with one or more of the individually administered general intelligence tests developed for that purpose.

SIGNIFICANTLY SUBAVERAGE is defined as IQ more than two standard deviations below the mean for the test.

ADAPTIVE BEHAVIOR is defined as the effectiveness or degree with which an individual meets the standards of personal independence and social responsibility expected for age and cultural group.

DEVELOPMENTAL PERIOD is defined as the period of time between birth and the 18th birthday.

Within the framework of the definition of mental retardation, an individual may meet the criterion of mental retardation at one time in life and not at some other time. A person may change status as a result of changes or alterations in his/her intellectual functioning, changes in his/her adaptive behaviors, changes in the expectations of the society, or for other known and unknown reasons. Decisions about whether an individual is classified as mentally retarded at any given time are always made in relation to behavioral standards and norms and in comparison to the individual’s own chronological age group.

The definition of mental retardation requires that an individual show deficiencies in both adaptive behavior and intellectual function. The same criterion is used to determine the degree or level of mental retardation. The levels of retardation are divided into four categories; mild, moderate, severe, and profound. The separation into four levels of mental retardation is scaled in terms of standard deviation units which describe the distribution of scores in the general population that would be expected for a particular test if the abilities measured by that test are normally distributed. These levels are not static and should only be one of several criteria used in planning a program for the individual who is retarded.

Adaptive behavior is a composite of many aspects of behavior and a function of a wide range of specific abilities and disabilities. Adaptive behavior is categorized in terms of degrees of impairment. These levels are scaled from mild negative deviations in adaptive behavior to almost complete lack of adaptation at the extreme lower limit. Changes in adaptive behavior may occur during the lifetime of the individual. These often occur through training or when marked changes are made in the environment. Deficits in adaptive behavior will also vary for different age groups. These variations can be seen in the following areas:
During Infancy and Early Childhood in:

1. Sensory-motor skills development.
2. Communication skills (including speech and language)
3. Self-help skills.
4. Socialization (development of ability to interact with others).

During Childhood and Early Adolescence in:

5. Application of basic academic skills in daily-life activities.
6. Application of appropriate reasoning and judgment in mastery of the environment.
7. Social skills (participation in group activities and interpersonal relationships).

During Late Adolescence and Adult Life in:

8. Vocational and social responsibilities and performance.

Resource Material:


Most authorities accept the figures of 3 percent of the population as being mentally retarded. This would mean that there are over four million retarded people in the United States alone. However, in an institutional setting, 40 to 50 percent of the residents are profoundly or severely retarded. In the general population, the severely and moderately retarded groups make up about 20 percent of the total group of retarded, which are about 4 in every 1000 people.
# Educational Terms and Degrees of Mental Retardation

<table>
<thead>
<tr>
<th>Educational Terms and degrees of Mental Retardation</th>
<th>Pre-School Age 0 – 5 years Maturation and Development</th>
<th>School Age 6 – 20 years Training and Education</th>
<th>Adult 21 years and over Social and Vocational Adequacy</th>
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<tr>
<td><strong>Profound</strong></td>
<td>Gross Retardation; minimal capacity for functioning in sensorimotor areas; need nursing care</td>
<td>Some motor development present; may respond to minimal or limited training in self-help.</td>
<td>Some motor and speech development; may achieve very limited self-care; needs nursing care.</td>
</tr>
<tr>
<td>“Life-support” level</td>
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<td>IQ = 19 and below</td>
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<td><strong>Severe</strong></td>
<td>Poor motor development; speech is minimal; generally unable to profit from training in self-help; little or no communication skills.</td>
<td>Can talk or learn to communicate; can be trained elemental health habits; profits from systematic habit training.</td>
<td>May contribute partially to self-maintenance under complete supervision can develop self-protection skills to a minimal useful level in controlled environment.</td>
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<tr>
<td>“Dependent Retarded” (custodial)</td>
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<tr>
<td><strong>Moderate</strong></td>
<td>Can talk or learn to communicate: poor social awareness; fair motor development; profits from training in self help; can be managed with moderate supervision.</td>
<td>Can profit from training in social and occupational skills; unlikely to progress beyond second grade level in academic subjects; may learn to travel alone in familiar places.</td>
<td>May achieve self-maintenance in unskilled or semi-skilled work under sheltered conditions; needs supervision and guidance when under mild social or economic stress.</td>
</tr>
<tr>
<td>“Trainable”</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>IQ = 36 to 51</td>
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<tr>
<td><strong>Mild</strong></td>
<td>Can develop social and communication skills minimal retardation in sensorimotor areas often not distinguished from normal until later age.</td>
<td>Can learn academic skills up to approximately sixth grade level by late teens. Can be guided toward social conformity.</td>
<td>Can usually achieve social and vocational skills adequate to minimum self-support but may need guidance and assistance when under unusual social economic stress.</td>
</tr>
<tr>
<td>“Educable”</td>
<td></td>
<td></td>
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<tr>
<td>IQ = 52 to 67</td>
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Information from An Introduction to Mental Retardation, Problems, Plans and Programs. The Secretary’s Committee on Mental Retardation, Washington, D.C., June 1965.
American Association on Mental Retardation (AAMR) developed the Manual on Terminology and Classifications of Mental Retardation. The purpose of the manual is to provide uniformity in terminology and is the medical and behavioral classifications of persons who are mentally retarded. Included in the study guide is a simplified classification of these etiologies. Specific etiologies are listed under each major classification.

The classification system is designed to furnish statistical information about the retarded individual. This system furnishes classificatory data on incidence, prevalence, characteristics, and concomitant information. Mental retardation is not a simple disease, syndrome, or symptom but is a state of impairment manifested by the behavior of the individual, and its causes are many. In most cases the specific cause of retardation is unknown. Clinical judgment is the measurement tool most widely used for differentiating diagnosis. Mental retardation may coexist with other handicaps and usually does with the more severely and profoundly retarded.

**Major Classifications Outline**

I. Infections and Intoxications – this classification includes maternal and child infectious diseases and intoxications.

   A. Prenatal Infections
      1. Cytomegalic Inclusion Body Disease
      2. Rubella
      3. Syphilis
      4. Toxoplasmosis

   B. Postnatal Infections – infections due to virus, bacteria, parasites, protozoa, and fungi. Example: meningitis and encephalitis.

   C. Prenatal Intoxications
      1. Toxemia of Pregnancy
      2. Alcoholism
      3. Drugs

   D. Postnatal Intoxications
      1. Lead Poisoning
II. Trauma or Physical Agents – cause associated with injury of the brain due to trauma, mechanical, or physical agents.
   A. Prenatal Injuries – X-ray and radiation
   B. Mechanical Injuries at Birth
   C. Anoxia at Birth; Perinatal Hypoxia

III. Metabolism or Nutrition
   A. Category includes disorders directly due to metabolic, nutritional, endocrine, or growth dysfunctions.
   B. Specific Disorders
      1. Tay-Sach’s Disease
      2. Hurler’s Disease (Syndrome)
      3. Galactosemia
      4. Phenylketonuria
      5. Thyroid Dysfunction – Cretinism
      6. Hyperuricemia (Lesch-Nyhan Syndrome)

IV. Gross Brain Disease
   A. Category includes neoplasms and a large group of heredo-generative disorders in which etiology is unknown or uncertain.
   B. Specific Disorders
      1. Von Recklinghausen’s Disease
      2. Sturge-Weber Disease
      3. Tuberous Sclerosis

V. Unknown Prenatal Influences
   A. Category includes:
      1. Conditions for which there is no definite etiology but which existed at or prior to birth.
      2. Primary cranial anomalies and congenital deficits.
   B. Specific Disorders
      1. Cornelia de Lange Syndrome
      2. Microcephaly
      3. Menigocele
      4. Myelomeningocele
      5. Hydrocephalus
      6. Spina Bifida
VI. Chromosomal Abnormalities

A. Category includes conditions associated with chromosomal aberrations.

B. Possible causes for such aberrations:
   1. Radiation
   2. Drugs and other chemicals
   3. Geno-mutations

C. Specific Disorders
   1. Cri-du-Chat Syndrome
   2. Trisomy 18
   3. Down’s Syndrome
   4. Klinefelter’s Syndrome
   5. Turner’s Syndrome

VII. Gestational Disorders – a number of defects are related to typical gestations.

A. Prematurity
B. Postmaturity

VIII. Psychiatric Disorders

A. Category includes retardation following psychosis or other psychiatric disorders where there is no evidence of cerebral pathology.

B. Specific Disorders
   1. Infantile Autism
   2. Childhood Schizophrenia

IX. Environmental Influences – adverse environmental conditions may cause mental retardation even when there is no evidence of organic disease or pathology.

A. Sensory Deprivation
B. Psychosocial Disadvantaged

X. Other Conditions – Classifications includes those situations where mental retardation is caused by defects in one or more of the special senses or where there appears to be multiple biological and social conditions contributing to slow or retarded development. It also includes illness-defined or unknown conditions.

A. Deficit of Special Senses – Conditions must be the only contributing factor to retardation Examples: Blindness, deafness, or combination of both.
MAJOR CLASSIFICATIONS SUBGROUPS

I. Intoxications and Infections

A. Prenatal Infections
   1. Major Diagnostic Features: Failure to thrive, mental retardation, microcephaly, chorioretinitis, and seizures.
   2. Mental Retardation: Varies from minimal to marked.
   3. Manifestations:
      a. General: Small or premature infant: Failure to thrive.
      b. Cardiac: Congestive Failure
      c. Skin: Petechiae, (pinpoint purple red spots caused by submucous hemorrhage) purpura, (bruises and hemorrhage beneath skin) and jaundice.
   4. Cause: Viral infections present in the salivary glands of the mother late in pregnancy and are passed through her circulatory system.
   5. Treatment: Many die soon after they are born. Those who survive usually have central nervous system damage. Seizures usually can be controlled with anticonvulsants.

B. Rubella (German Measles)
   1. Major Diagnostic Features: Congenital heart disease, deafness, cataracts, or glaucoma, and mental/motor retardation.
   2. Mental Retardation: Ranges from no involvement to profound.
   a. Manifestations:
      (1) General: Prematurity, low birth weight, and failure to thrive. The degree of organ and tissue involvement varies greatly.
      (2) Eyes: Unilateral or bilateral nuclear cataracts or congenital glaucoma
      (3) Ears: Nerve deafness of varying degree.
      (4) Cardiac: Involvement can vary from mild to severe.
      (5) Skin: A red-purplish macular rash and jaundice during newborn period.
   3. Cause: Most devastating when mother contracts rubella during first trimester of pregnancy.
   4. Treatment: Surgery should be done on correctable lesions.

C. Syphilis
   1. Major Diagnostic Features: Rash, hepatosplenomegaly, anemia, jaundice, and ostochondritis are early manifestations. Late manifestations include condylomas, (wart-like lesions of the skin) gummas, deafness, saber shins, and central nervous system involvement.
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PTEC 155 – Developmental Disabilities

Module 27 – Etiologies

2. Mental Retardation: Level of retardation varies depending upon the degree of central nervous system damage.

3. Manifestations:
   a. General: There are two types – early and late congenital syphilis.
   b. Nose: Rhinitis or snuffles with nasal obstruction. Small, saddle shaped nose.
   c. Mouth: Peg-shaped permanent teeth. Perforation of the palate (late).
   d. Laboratory Findings: Positive serology.

4. Cause: Spirochetal (venereal) infection in mother that is not treated.

5. Treatment: Penicillin is the drug of choice.

D. Toxoplasmosis

1. Major Diagnostic Features: Two clinical pictures may be caused by infection of the fetus – hydrocephalus, chorioretinitis, intercranial calcifications, and mental retardation representing more prolonged infection; acute hemolytic anemia, jaundice, purpuric rash, and hepatosplenomegaly representing more recent infection.

2. Mental Retardation: Varies in degree from mild to severe.

3. Manifestations:
   a. General: Prematurity
   b. Head: Hydrocephalus or microcephalus
   c. Skin: Purplish rash at birth
   e. Laboratory Findings: Parasites isolated from the cerebrospinal fluid. Positive skin test in the infant.


5. Treatment: Because of the high mortality rate, treatment with pyrimethamine and sulfadiazine should be instituted.

6. Postnatal Infections: In the past, a relatively large percentage of mental retardation was caused by infections occurring in and around the brain postnatally. These clients often have mental retardation, spasticity, paralysis, and seizures.
Module 27 – Etiologies

a. Bacterial Intracranial Infections: Meningitis can be caused by different bacteria or virus. The common causes of bacterial meningitis are meningococci and pneumococci. The advent of antibiotics has lessened the incidence of meningitis and, hence, the incidence of mental retardation from this cause is also less.

b. Viral Infections: The problem of viral infections in and around the brain presents a different picture. There are no drugs available to treat viral infections. Vaccines that have been developed for rubella, mumps, and other viruses that have aided in prevention of these diseases can reduce the incidence of retardation. The signs and symptoms in a person with retardation following viral encephalitis are similar to those following bacterial meningitis.

7. Prenatal Intoxications

a. Toxemia of Pregnancy: The symptoms manifested by the mother include hypertension, edema, fluid retention, albuminuria, and decreased urinary output. Occurrence is usually in the last trimester of pregnancy. In severe or untreated cases, mental retardation may occur in the newborn. These children show symptoms of generalized brain damage that can vary from borderline to severe.

b. Alcoholism: Currently, alcoholism is clearly related to a “pattern of craniofacial, limb, and cardiovascular defects associated with prenatal onset growth deficiency, and developmental delay.” This pattern has become known as the fetal alcohol syndrome.

The signs of fetal alcohol syndrome can range from slight retardations that are not detected until much later when developmental problems arise to severe retardation with extensive brain stem involvement. Some of the defects and deficiencies observed are: low birth weight, microcephaly, craniofacies abnormalities, cardiac anomalies, and fine motor dysfunction.

c. Drugs: The Thalidomide tragedy of 1960 to 1962 increased awareness of the potential problems related to birth defects from prescription and nonprescription drugs. Very little is known that can be applied to all pregnancies, but the greatest danger of inducing malformations is in the first trimester of pregnancy. Since the effects of many drugs have not been studied, a general rule of thumb is to avoid all medications during pregnancy, especially during the first trimester. (Vitamin and mineral supplements are exceptions.)
The effects of certain drugs on the fetus or neonate are:

<table>
<thead>
<tr>
<th>Maternal Medications</th>
<th>Clinical Manifestations</th>
</tr>
</thead>
<tbody>
<tr>
<td>(1) Salicylates (large amounts)</td>
<td>(a) Neonatal Bleeding</td>
</tr>
<tr>
<td>(2) Streptomycin</td>
<td>(b) Possible VIII (eighth) cranial nerve deafness</td>
</tr>
<tr>
<td>(3) Erythromycin</td>
<td>(c) Liver damage</td>
</tr>
<tr>
<td>(4) Heroin and Morphine</td>
<td>(d) Increased neonatal death</td>
</tr>
<tr>
<td>(5) Phenobarbital (in excess)</td>
<td>(e) Neonatal Bleeding; death</td>
</tr>
<tr>
<td>(6) Smoking</td>
<td>(f) Low birth weight</td>
</tr>
</tbody>
</table>

d. Maternal-Fetal Blood Group Incompatibilities: Mental retardation secondary to kernicterus due to Rh factor and A, B, O, incompatibilities has been reported.

(1) Major Diagnostic Features: Elevated bilirubin, jaundice, and brain damage secondary to overproduction of unconjugated bilirubin; mental retardation varies from minimal to severe.

(2) Resultant condition known as erythroblastosis fetalis.

(3) Manifestations:
   (a) Eyes: Nystagmus
   (b) Gastrointestinal: Poor feeding and vomiting during acute stage
   (c) Skin: Jaundiced

(4) Prevention and Treatment: Exchange transfusion during the newborn period when indicated usual prevents kernicterus. In severe cases intrauterine transfusions have been done.

e. Postnatal Intoxications

Lead Poisoning: The ingestion of lead by infants chewing on beds, toys, or other objects painted with lead-based paints has in the past been reported as producing retardation. Lead poisoning is relatively rare today due to the recognition of the danger and the precautions. Most paints on the market are not lead based.

Lead causes a degeneration of the peripheral nerve cells, principally motor nerves, causing motor paralysis. Degeneration occurs first in the radial nerve producing wrist drop and paralysis of the extensors of the fingers. If exposure continues, there will be a atrophy of the muscles with generalized paralysis and spasm of many muscle groups.
Degeneration of cortical cells leads to retardation in some cases. Lead is characterized by a blue line around the gum tissue.

II. Trauma and Physical Agents

A. Prenatal Injuries:

It was once thought that the fetus was invulnerable to trauma. This has been proven to be untrue.

Trauma, such as a blow to the pregnant woman’s abdomen during the third trimester can sometimes cause injury to the fetus. Some infants are born dead or die shortly after birth. Postmortem examinations find subdural hematomas, tears of the brain, or hemorrhages into the brain substance.

In infants that survived, some were mentally retarded. The degree of retardation varied from near normal to severe depending on the extent of the trauma. Some had motor impairment in varying degrees.

X-ray and Radiation:

Studies have been done to determine the incidence of mental retardation and exposure to x-ray and radiation. Many believe that the greatest danger may be the exposure of the ovaries and/or testes to radiation producing chromosome changes, which may result in abnormalities in the fetus. Many abnormalities have been attributed to this exposure in humans.

X-ray exposure during the first trimester has produced abnormalities due to the interruption of cell division.

B. Mechanical Injury at Birth:

Mechanical injury at birth can be the result of several factors. The degree of retardation and the other signs and symptoms depend entirely on the type and extent of damage and the involved area of the brain. There may be localized damage producing paralysis of one hand, one arm, one leg, etc. Aphasics may develop due to localized brain damage. The damage may be generalized and produce more massive signs and symptoms such as generalized spasticity, rigidity, or flaccidity. There may be no muscular involvement, but the infant may have varying degrees of retardation. Forces that produce mechanical damage include improper application of forceps, birth canal compression of the head, prolonged labor, malpositions producing long labors, head-pelvic disproportions, etc. These may produce either tears to the brain or hemorrhages. Good obstetrical care, better prenatal care, and greater awareness of the problems have contributed to the reduction in incidence or retardation from these causes.
C. Anoxia At Birth: *(Perinatal hypoxia)*
Encephalopathy secondary to anoxia or asphyxia at birth has also been reduced substantially as an etiological agent in mental retardation recent years. Many of the same factors that can produce mechanical injury can also produce asphyxia. This may be due to delayed respiration; compression, knotting or twisting of the umbilical cord to premature separation of the placenta; and many other forces preventing proper oxygenation of the fetal blood.

“Hold-Back syndrome” where the head prevented manually from being delivered. During which time the placenta is separating and fetal placental circulation is decreasing in efficiency, can produce anoxia with permanent brain damage. The longer the period of delay, the greater the damage that occurs. These conditions may all produce similar types of generalized brain damage in which both cortical and deep structures are involved. The degree of retardation can vary depending on the length of time the infant is deprived of oxygen.

D. Postnatal Anoxia:
Any condition that inhibits or decreases the supply of oxygen to the brain can cause mental retardation that varies in severity.

Some conditions that can cause anoxia in the small child are: *near drowning, choking on objects, and electrocution.*

E. Postnatal Injury:

Direct trauma to the head, which produces brain tissue, tears and hemorrhages, with or without multiple skull fractures, may cause retardation. The degree of retardation and physical handicaps depends on the area of the brain involved and the extent of the damage.

Automobile accidents, falls, accidental shootings, and bicycle accidents are just some of the causes. Any severe blow to the head may cause brain damage.

**BATTERED CHILD SYNDROME**

The term “Battered-Child Syndrome” has been used to refer to the results of emotional and nutritional neglect as well as physical abuse.

Most often, battered children are toddlers or preschool-age, although infants and teenagers have also been victims of abuse. Usually, the child’s general health and hygiene are poor, malnutrition is evident, and the injuries are both old and new, children suffering from chronic neglect are usually subnormal in both height and weight.

Retardation in intellectual and social development is not uncommon. Brain damage is usually the result of subdural hematomas. Evidence of this trauma may be skull fractures. Level of retardation can vary mild to severe.
III. Metabolism and Nutrition

A. Introduction

In recent years, much emphasis has been placed on discovery and understanding of enzymatic and carrier defects; many of which are associated with mental retardation of varying degrees. Although these biochemical lesions do not account for a very large proportion of the mentally retarded, their early discovery and treatment may, in many cases, result in clinical recovery and in prevention of central nervous system deterioration.

B. Specific Disorders:

1. Tay-Sach’s Disease: (Amaurotic Familial Idiocy) A disorder of lipid metabolism.
   a. Physical Features:
      (1) HEAD: Initially, the head size is normal. Those who survive two years or longer have generalized head enlargement.
      (2) EYES: A cherry-red macular spot can be detected at about 2 1/2 months of age.
      (3) NERVOUS SYSTEM: Motor development, normal until five months of age. From the sixth month on, there is a progressive failure to acquire new skills and loss of previously acquired skills. Frequent findings are exaggerated motor response to sound and excessive drooling. There are focal or generalized convulsions after the first year. After the first two years, spasticity and rigidity are prevalent.

   b. Treatment and Prognosis:

      The disease is progressive, with most children dying between two and four years of age. Death is often the result of aspiration pneumonia.

   c. Genetics:

      Autosomal Recessive (Autosome: any of the paired chromosomes, other than the sex x and y chromosome.) Found most frequently in Jewish families. Two parents each of which carries the recessive gene.
Hurler’s Syndrome:

- Synonyms: Gargoyleism, Mucopolysaccharoidosis, Lipochondrodystrophy (Abnormality in the skeletal bones and cartilage).

  Characterized by faulty starch and sugar metabolism.

- Major Diagnostic Features:
  1. Mental Retardation
  2. Course features
  3. Skeletal abnormalities
  4. Hirsutism (A condition characterized by the excessive growth of hair, or the presence of hair in unusual places.)
  5. Hepatomegaly (enlarged liver)
  6. Characteristic facial appearance

- Inheritance and Etiology
  1. Autosomal Recessive
  2. Accumulation of mucopolysaccharides in tissue

- Mental Retardation:

  Severe and usually present by the first year of life.

- Clinical Characteristics:
  1. Head:
     Large, prominent forehead, frequently scaphocephalic (boat shaped head)
  2. Eyes:
     Corneal clouding, hypertelorism, thick eyelids, coarse brows and lashes
  3. Ears:
     Low set; hearing loss
  4. Nose:
     Wide tip and nostrils, flat bridge, frequent rhinitis
  5. Cardiac:
     Enlarged: frequent congestive failure
  6. Gastrointestinal:
     Protruberant abdomen, umbilical hernia, hepatosplenomegaly
  7. Mouth:
     Open with large protruding tongue, thick lips, high and narrow palate. Small peg-shaped teeth.
3. **Galactosemia:**
   This metabolic error impairs the client's ability to metabolize galactose found in milk.
   
   a. **Physical Features:**
      (1) **Eyes:**
          In the untreated client, cataracts develop within the first year.
      (2) **Abdomen:**
          Almost all untreated clients develop hepatomegaly.
      (3) **Skin:**
          In many untreated infants, jaundice develops in the first or second week of life.

   b. **Nervous System:**
      Untreated infants are lethargic and hypotonic.

   c. **Treatment and Prognosis:**
      (1) Untreated clients have a high death rate in infancy, and those who survive are almost always retarded.
      (2) Treatment consists of dietary restrictions of galactose. If treatment is instigated early enough, the child may develop normally. Dietary restrictions continued for life, but it should be strictly adhered to until the child is more than four years old.

   d. **Genetics:**
      Autosomal recessive.

4. **Phenylketonuria:**
   Characterized by faulty protein metabolism
   
   a. **Synonym:** PKU

   b. **Major Diagnostic Features:**
      (1) Mental Retardation
      (2) Fair hair and skin
      (3) Eczema
      (4) Neurological abnormalities
      (5) Elevated blood levels of phenylalanine and its metabolites

   c. **Inheritance and Etiology:**
Due to the absence of phenylalanine hydroxylase, which converts phenylalanine to tyrosine, phenylalanine accumulates in the blood, and the deamination produces phenylpyruvic acid which is excreted in the urine. Damage to brain occurs by six months. Autosomal recessive mode of inheritance.

d. Mental Retardation:

Majority have moderate to severe retardation. If treated early, intelligence level may be increased.

e. Clinical characteristics:
   (1) General: Initially normal, but early onset of mental deterioration.
       Short stature, musty or mousy odor.
   (2) Head: Microcephaly
   (3) Eyes: Blue
   (4) Gastrointestinal: Vomiting in infancy
   (5) Neurological: 20 percent have convulsions. Agitated and restless behavior; hyperactive deep tendon reflexes. Hypotonia, fine rapid tremors, athetoid or jerky aimless movements.

f. Diagnostic Tests:
   (1) Electroencephalogram: Hypsarrhythmia (Abnormal height in wave)
   (2) GUTHRIE TEST: The inhibition assay is based on the growth requirements of a specific strain of Bacillus subtilis for phenylalanine. An inhibitor is used in the culture media. However, if phenylalanine is supplied in excess by filter paper saturated with PKU blood, bacterial growth will ensue.

Ferric chloride crystals turn bright green in presence of phenylpyruvic acid. Applied to diaper of newborns and again at six-week checkups.

g. Prognosis
   (1) Untreated: 2-3 years, 85% have severe mental retardation.
   (2) Treated: Phenylalanine-restricted diet started in infancy. Lofenelac or Ketonil are the milk substitutes that these infants ingest in place of milk. Close to normal intelligence.
5. Thyroid Dysfunction: (Hypothyroidism, Congenital Cretinism)
   a. Major Diagnostic Features:
      Mental retardation in varying degrees, delayed skeletal
      maturation, drug and cold extremities, myxedema, and
      characteristic facial appearance with large protruding tongue,
      swollen lips and nose, facial edema of non-pitting type.
   b. Manifestations:
      (1) Head: The head appears large
      (2) Eyes: Appear wide-set
      (3) Nose: Broad bridge
      (4) Neck: Short and thick
      (5) Gastrointestinal: Prominent abdomen with an umbilical
         hernia; feeds poorly; constipation.
      (6) Skeletal: Short extremities, broad hands with short fingers.
         Shortness of stature, is often severe.
   c. Recognized Forms:
      (1) Familial Cretin:
         Can usually be diagnosed by history of other cretins in
         family.
      (2) Sporadic Cretin
         Due to failure of the thyroid gland to develop in fetal state.
      (3) Goiterous Cretin:
         Caused by a thyroid goiter
      (4) Infinite Cretinism:
         A condition, which results from thyroid deficiency in fetal or
         early neonatal life
   d. Treatment and Prognosis:
      In general, adequate replacement of thyroid hormone results in
      reversal of the signs and symptoms.

6. Hyperuricemia with Mental Retardation:
   a. Synonyms:
      Lesch-Nyhan Syndrome the “Biting Sickness”
   b. Major Diagnostic Features:
      (1) Mental retardation
      (2) Self-Mutilation
      (3) Chorea-Athetosis
      (4) Hyperuricemia
c. Inheritance and Etiology:
   (1) X chromosome – linked recessive type of inheritance
   (2) Deficiency of hypoxanthinegranine phosphoribosyltransferase

d. Mental Retardation: Severe

e. Clinical Characteristics:
   (1) General:
       Normal appearing at birth - Irritable by two months. By two years, self-mutilation is present, manifested by lip-biting, finger-chewing, teeth grinding, and marked swinging of the arms.
   (2) HEENT (Heart, Eyes, Ears, Nose, Throat):
       Lesions of lips secondary to lip-biting
   (3) Renal:
       Urine pH is acid. Red blood cells and renal damage secondary to uric acid stones.
   (4) Skeletal:
       Deformities of the hands secondary to finger-chewing.
   (5) Central Nervous System:
       Slow motor development, spasticity, choreo-athetosis follows. Muscle hypertrophy secondary to torsion spasms.

f. Diagnosis:
   Increased uric acid levels in the blood and urine.

g. Prognosis:
   Death is secondary to severe renal or neurological damage

h. Treatment:
   Allopurinol and Benemid decrease uric acid level and also yield some clinical improvements.

IV. Gross Brain Disease

Specific Disorders

A. Von Recklinghausen’s Disease (Neurofibromatosis, Elephant Man’s Disease)

1. Physical Features:

   a. Skin: Patches of increased pigmentation (café-au-lait spots) often present at birth. Skin tumors appear in late childhood or early adolescence. Scoliosis of the back is frequent.
   b. Multiple neurofibromata occurring on the skin along the course of the nerves; associated with marked cutaneous pigmentation.
2. Nervous System:
   a. Mental deficiency is a common finding but not present in all cases.

3. Treatment and Prognosis:
   a. Surgical removal of tumors is recommended when they are
   b. Disfiguring or cause mechanical damage.

4. Genetic:
   a. Autosomal recessive

B. Sturge Weber Dimitri

1. Synonyms:
   a. Cerebral Angiomatosis, Encephalotrigeminal Angiomatosis,
   b. Encephalofacial Angiomatosis

2. Major Diagnostic Features:
   a. Venous Angioma: Trigeminal in distribution, involving the skin, scalp, skull, and meninges.
   b. Seizures and other neurological abnormalities
   c. Mental retardation.

3. Inheritance and Etiology:
   a. Familial, but genetic transmission not clear

4. Mental Retardation:
   a. Varies from normal intelligence to severe retardation depending upon the secondary neurological damage.

5. Clinical Characteristics:
   a. Head: Asymmetry may occur
   b. Eyes: Glaucoma and buphthalmos (childhood glaucoma), may occur on the side of the lesion; hemianopia (blindness for one half field of vision), and choroidal angiomas are frequently found.
   c. Mouth: Port-wine staining of the buccal mucosa
d. Skin: Nevus flammeus (port-wine stain) is usually found at birth on the same side as the cerebral angiomatosis. Nevus or angiomatosis involves the forehead and cheek and is trigeminal in distribution, although it can extend across the midline to the other side or other areas of the skin.

e. Skeletal: The nevus may involve bone with secondary hypertrophy.

f. Central Nervous System: The vascular abnormality or the underlying cortical defect may cause hemiplegia. Seizures may be unilateral or become generalized. Calcium deposits are found in the layers of the cortex.

6. Prognosis:

a. Convulsions are often difficult to control. Hemispherectomy beneficial in rare cases.

C. Tuberous Sclerosis

1. Synonyms:
   (1) Adenoma Sebaceum
   (2) Bourneville-Pringle Syndrome
   (3) Epiloia

2. Major Diagnostic Features:
   (1) Seizures
   (2) Adenoma sebaceum
   (3) Mental retardation

3. Inheritance and Etiology:

a. In some instances, the condition appears to be an autosomal dominant; in others a recessive type of inheritance.

4. Mental Retardation:

a. Varies in severity but usually quite severe with increased neurological involvement.

5. Clinical Characteristics:

a. Facies: Pink to red-orange nodules in butterfly distribution – all of face may be involved.

b. Eyes:
Pupils may be unequal secondary to brain involvement.

c. Mouth:
   Fibromas of gums often present.

d. Skin:
   Shagreen patch on trunk (elevated area with abnormal pigmentation, orange-peel appearance, fibromas of nails, forehead, scalp, graying hair, café-au-lait spots, hemangiomas, oral areas of depigmentation in infants with seizures.

e. Central Nervous System:
   Seizures may occur early in life secondary to mass in central nervous system.

f. Other:
   Tumors of kidneys, heart, eyes, lungs, spleen.

6. Diagnostic Tests:

   a. Abnormal EEG; intracranial calcification on x-ray films of the skull; abnormal pneumoencephalogram, angiogram and ventriculogram depending upon the location and extent of brain lesion.

7. Treatment:

   a. Control of seizures, although this may be difficult.

V. Unknown Prenatal Influences

Specific Disorders

A. de Lange’s Syndrome

1. Cornelia de Lange syndrome
2. Amsterdam Dwarf

   a. Major Diagnostic Features:
      (1) Bushy, confluent eyebrows
      (2) Upturned nose
      (3) Wide upper lip
      (4) Hirsutism and Skeletal abnormalities
      (5) Small for age infants

   b. Inheritance and Etiology:
      (1) The exact etiology is unknown, although there is probably a genetic component.

   c. Mental Retardation: Severe

   d. Clinical Characteristics:
      (1) General:
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Below third percentile for both height and weight. Low pitched growling cry.

(2) Head:
Microbrachycephalic (Having a head disproportionately short).

(3) Eyes:
Antimongoloid slant; pallor of optic disks, ptosis (dropping of the upper eyelid), nystagmus (constant, more or less cyclical movement of the eyeball, in any direction), bushy, confluent eyebrows; long curly eyelashes.

(4) Nose:
Small upturned with anteverted nostrils (included or bent forward).

(5) Mouth:
Fish-like with a thin, wide upper lip, palate is narrow and high arched, widely spaced teeth, micrognathia (abnormally small jaws) is frequent.

(6) Skeletal:
Micromelia, phocomelia, oligodactyly (subnormal number of fingers and toes, clinodactyly (permanent deflection of one or more fingers), proximally placed thumb, short tapered fingers, decreased thenar eminence (fleshy pad at base of thumb), flat spade-like hands, flexion contractures of elbow, and short toes with syndactyly (fusion of two or more toes or fingers) of second and third toes.

(7) Skin:
Extreme hirsutism, cutis marmorata (purplish discoloration of skin, on exposure to cold), dusky hue of face, hypoplastic (defective development) nipples, deficient epidermal ridge patterns and a simian line.

e. Prognosis:

(1) Poor, death is usually due to respiratory infection or aspiration pneumonia.

B. Microcephaly:

1. Major diagnostic features:

a. An abnormally small head which is most often associated with mental retardation of variable degrees.
2. Inheritance and etiology:
   a. There are numerous causes of microcephaly. The familial type is autosomal recessively inherited. Other causes are toxoplasmosis, intrauterine irradiation, cranial synostosis (fusion of adjacent bones), etc. Most common cause is cerebral anoxia and/or nonspecific.

3. Mental Retardation:
   a. Usually moderate to profound in familial type of microcephaly.

4. Clinical Characteristics:
   a. Facies: Normal size
   b. Head: Criteria for diagnosis:
      (1) Head circumference less than 5 centimeters below expected average during early childhood.
      (2) Head circumference below 43 centimeters at maturity.
      (3) Brain weight under 1000 Grams at maturity.
   c. Ears:
      (1) Relatively large and protruding in familial type
   d. Nose:
      (1) Narrow or flat bridge; relatively large
   e. Mouth:
      (1) High-arched palate in familial type
   f. Central Nervous System: Retarded mental, motor development; hypertonia

5. Treatment:
   a. Special schooling

C. Spina Bifida: (See Figure 11 – 11)
   1. Congenital defect in walls of spinal canal caused by lack of union between the laminae of the vertebrae. Lumbar portion is part chiefly affected. (Results in tumors varying in size from walnut to size of a child’s head. Tumors are defined as the following items 4, 5)

D. Meningocele: (See Figure 11 – 11)
   1. A meningocele is a herniation of the meningeal sac of nervous system (skull or spinal column) through a bony defect (spina-bifida) – but not through the skin. The nerves are not disturbed.
E. Myelomeningocele:  *(See Figure 11–11)*

1. Congenital defect *(spina bifida)* with portion of the cord and membrane (meninges) protruding through the vertebra. The skin is thinned out, the nerves are in the sac, and there is a paralysis below the sac. The degree of paralysis depends upon the disturbance of the innervation of the nerves. Often occurs in the lumbar area.

F. Spina Bifida Occulta: *(See Figure 11–11)*

1. Failure of vertebrae to close but lacking hernia protrusion.

G. Hydrocephalus:

1. Major Diagnostic Features:
   a. Increased head size
   b. Protruding eyes with deficiency in upward gaze. *(Sundown eyes)*
   c. Poor motor development
   d. Neurological abnormalities with mental retardation

2. Inheritance and Etiology:

   a. Usually no genetic basis but x-linked stenosis of aqueduct of Sylvius has been reported.
      (1) Acquired:
         Infections (toxoplasmosis, meningitis, etc.)
      (2) Congenital:
         Results from obstruction of some part in the cerebrospinal fluid pathway.
         (a) Communicating
            (i) Cerebral aqueduct of Sylvius
            (ii) Foramina of exit of fourth ventricle
         (b) Noncommunicating
            (i) Cistema of basilar meninges
PHOTO COPY OF DIAGRAM FROM PAGE 36 GOES HERE
3. Mental Retardation:
   a. Varies depending upon the location and amount of obstruction.

4. Clinical Characteristics:
   a. Head:
      Unusually normal or slightly enlarged at birth and then increases at a greater than normal rate Fontanel is tense and tends not to close. Sutures remain open and scalp veins are prominent.
   b. Eyes:
      Roofs of orbits depressed pushing eyes downward and outward. Optic atrophy, limitation of upward gaze, retraction of upper lids, and strabismus are frequently seen.
   c. Skeletal:
      Various types of deformities have been noted.
   d. Central Nervous System:
      Sixth nerve palsies, hyperactive deep tendon reflexes, spasticity, extensor plantar response, and seizures are frequently encountered. Also found are middle cranial and spinal defects with meningocele or menigomyocele and Arnold-Chiari (A condition in which there is displacement of the medulla and cerebellum into the opening in the basilar part of the occipital bone. It is usually accompanied by spina bifida, myelomeningocele and seizures) syndrome. Cortical convolutions are flattened, ventricular system is dilated secondary to obstruction.

5. Prognosis:
   a. Occasionally arrests spontaneously; usually progresses to mental retardation, spasticity, blindness, and death during infancy or early childhood.

6. Treatment
   a. Placing a tube to drain cerebrospinal fluid ventricles of brain to heart chamber, peritoneum, or kidneys (shunting procedure). Conscientious client care with attention to prevention of pressure sores, respiratory complications, and maintenance of the neuromuscular system.
b. Majority (95%) of these clients are trisomic for chromosome #21 due to nondisjuntion (failure of chromosomes to separate). Greatly increased risk for repeat Down’s, if first Down’s is translocation type (Chromosomal aberration in which a segment from one chromosome becomes united to another of different type). Male Down’s not known to reproduce. Female Down’s have 50/50 risk for Down’s infant with each pregnancy. Risk of having Down’s infant is higher when any mother-to-be is 40-years-old.

7. Mental Retardation: IQ beyond infancy usually less than 50. May range considerably. Mosaics may be only mildly retarded.

8. Clinical Characteristics:

a. Head, eyes, ears, nose throat: Brachycephaly (Anterior posterior shortening, with increased height of skull), with a flattened occiput. Broad short nose. Eyes have epicanthal fold. Oblique palpebral (eyelid) fissures. Brushfield Spots (brownish flecks in the iris), strabismus (crossed eyes), and nystagmus. Ears are malformed with an angular, overlapping helix and a prominent antihelix. Neck is broad and short with excessive loose skin over nape. Mouth is held open, showing a protruding tongue, small teeth, high arched palate, fissured lips and furrowed tongue, and occasionally a cleft lip and palate.

b. Cardiac: 20 percent have congenital heart disease with endocardial cushion defect being most common

c. Skin: Simian line, with abnormal dermatoglyphics, single flexion crease of the fifth finger

d. Skeletal: Hands are short and broad. Incurved fifth finger, increased space between first and second toes, plus a plantar furrow.

e. Treatment: Correction of congenital malformations should be considered as the need arises. The most frequent causes of death are respiratory infections and congenital heart disease. Early treatment with antibiotics is beneficial.

H. Klinefelter’s Syndrome:

1. Major Diagnostic Features:

a. Tall in stature

b. Decreased secondary sexual characteristics
2. Mental Retardation:
   a. When present, it is usually mild

3. Inheritance and Etiology
   a. Nondisjunction results in XXY type sex chromosomes. Rare XXXY and XXXXY as well as mosaic (XXY, XXXY, etc.)

4. Clinical Characteristics:
   a. General:
      Character of personality disorders common
   b. Head:
      Frontal hyperostosis (excessive growth of bony tissue)
   c. Eyes: Myopia and Astigmatism
   d. Mouth: Cleft palate, marked mandibular overgrowth in clients with XXXXY
   e. Genitourinary:
      Small, firm testes, varying degrees of eunuchoidism, (resembling a castrated male). Azospermia (absence of spermatozoa in the semen), progressive fibrosis and hyalinization of the seminiferous tubules, decreased Leydig cell function (cells of the testis, which furnish its internal secretion), penis is normal size although hypospadias (anomaly in the male, in which the urethra opens on the under side of the penis or on the perineum) is present in clients with XXXY, XXXXY; decreased secondary sexual characteristics, and gynecomastia (over development of the mammary tissue in males).
   f. Skin:
      Diminished facial and body hair
   g. Skeletal:
      Tall and slender, frequently obese, lower limbs maybe larger than upper, radial ulnar synostosis (normal or abnormal union of two bones by osseous material), scoliosis, clinodactyly (permanent deviation or flexion of one or more fingers), and chest deformities.
   h. Diagnosis:
      Elevated urinary gonadotropin excretion. Low or normal ketosteroid levels. Positive sex-chromatin pattern
   i. Treatment: Hormonal therapy
VI. Chromosomal Abnormalities

Specific Disorders

A. Synonyms: Cry of Cat Syndrome

1. Major Diagnostic Features:
   a. Severe mental/motor retardation
   b. Microcephaly
   c. Rounded face with hypertelorism (Abnormal width between the eyes).
   d. Cat-like cry

2. Inheritance and Etiology
   a. Karyotype demonstrates deletion of part of the chromosomal material on one of the short arms of the 4–5 group. The possibility of recurrence in another member of the family is rare unless condition is due to a translocation chromosome.

3. Mental Retardation: Severe

4. Clinical Characteristics:
   1. General: Usually small for date infant, failure to thrive, abnormal cry is present at birth and persists until five months of age or longer.
   2. Facies: Rounded
   3. Head: Microcephalic
   4. Eyes: Hypertelorism, antimongoloid slant, epicanthal folds, strabismus, and optic atrophy.
   5. Ears: Low set and may be prominent.
   6. Mouth: Micrognathia (Abnormally small jaws), and occasionally cleft palate.
   7. Cardiac: Various types of congenital defects.
   8. Skin: Abnormal dermatoglyphics (surface markings on the skin)
   9. Central Nervous System: Hypotonia

5. Treatment: Usually institutionalization

B. Trisomy 18:

1. Major diagnostic features:
   a. Small for date infants with severe motor/mental retardation
   b. Overlapping of the index finger over the third finger
   c. Grecian nose; low-set ears, cleft lip and palate
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2. Manifestations:
   a. General: Infants usually do not survive beyond first year of life
   b. Head: Microcephalic with a prominent occiput.
   c. Other Findings: Congenital heart disease and renal abnormalities

3. Genetics:
   a. Caused by nondisjuntion (failure to separate) of one of the chromosomes in the 17 – 18 group. Possibility of this recurring in the same family is rare.

4. Treatment:
   a. Limited treatment because majority die within the first year.

C. Down’s Syndrome:

1. Synonyms:
   a. Mongolism
   b. Trisomy 21

2. Major Diagnostic Features:
   a. Typical facial appearance
   b. Mental retardation
   c. Muscular hypotonia
   d. Congenital Heart Disease

3. Inheritance and Etiology:
   a. Majority (95%) of these clients are trisomic for chromosome #21 due to nondisjunction (failure of chromosomes to separate). Greatly increased risk for repeat Down’s, if first Down’s is translocation type (Chromosomal aberration in which a segment from one chromosome becomes united to another of different type). Male Down’s not known to reproduce. Female Down’s have 50/50 risk for Down’s infant with each pregnancy. Risk of having Down’s infant is higher when any mother-to-be is 40 years old.

4. Mental Retardation:
   a. IQ beyond infancy usually less than 50. May range considerably. Mosaics may be only mildly retarded.
5. Clinical Characteristics:
   a. Head, Eyes, Ears, Nose, Throat: Brachycephaly (Anterior posterior shortening, with increased height of skull), with a flattened occiput. Broad short nose. Eyes have epicanthal fold. Oblique palpebral (eyelid) fissures. Brushfield Spots (brownish flecks in the iris), strabismus (crossed eyes), and nystagmus. Ears are malformed with an angular, overlapping helix and a prominent antihelix. Neck is broad and short with excessive loose skin over nape. Mouth is held open, showing a protruding tongue, small teeth, high arched palate, fissured lips and furrowed tongue, and occasionally a cleft lip and palate.
   b. Cardiac: 20 percent have congenital heart disease with endocardial cushion defect being most common.
   c. Skin: Simian line, with abnormal dermatoglyphics, single flexion crease of the fifth finger.
   d. Skeletal: Hands are short and broad. Incurved fifth finger, increased space between first and second toes, plus a plantar furrow.
   e. Treatment: Correction of congenital malformations should be considered as the need arises. The most frequent causes of death are respiratory infections and congenital heart disease. Early treatment with antibiotics is beneficial.

D. Turner’s Syndrome:

1. Major Diagnostic Features:

   a. Short stature
   b. Webbing and/or shortening of the neck
   c. Absence of secondary sex characteristics
   d. Typical facial appearance including a broad bridge of the nose, epicanthal folds, and low-set ears
   e. Mental retardation is not always present

VIII. Psychiatric Disorders

Specific Disorders

A. Childhood Schizophrenia:

1. Childhood schizophrenia is an over-generalized term used to describe two psychotic states. The term “schizophrenia” was probably employed to distinguish the fact that these psychotic states are “cognitive” disorders as opposed to “affective”, or mood disorders. The two states of psychosis observed in children are early infantile autism, and the symbiotic psychoses. The similarity of symptoms of the autistic child and the child
experiencing symbiotic psychosis, includes bizarre use of symbolism in communication. These children all demonstrate poor ability to function at comparable levels to “normal” peers and have a highly active fantasy life.

B. Symbiotic Psychosis

1. Where early infantile autism represents a failure of the attachment to the mothering figure, symbiotic psychosis is an over-attachment to the mother. Failure of individuation yields a state where the child feels annihilated by the absence of the mother. (Similar to, but more severe than the separation anxiety, experienced by normal toddlers.) Most of these children are highly resistant to change and are extremely dependent persons. In some cases, the pathology is promoted by an immature, overly conscientious, overly protective mother, who may have a vested interest in keeping the child dependent upon her. These children develop emotionally and cognitively as an extension of the mother. They may later in life, remain attached to mother symbols, such as the home, and fight to remaining the symbols presence. Due to the extreme anxiety these children, upon separation from the mother or mother symbol, experience and infantile or regressed state. Individuation, which might lead to separation from the mother, is resisted, hence the child may appear “slow” to learn and retarded due to lack of skills.

C. Infantile Autism

1. This diagnosis refers to children whose behavior is characteristically altered from the time they are born. Often parents state, “he/she did not respond with the usual social smiles and noises”, or “they are hard to love”.

2. Etiology of the autistic child has not been completely determined. Kanner’s criteria for the diagnosis of infantile autism are:

   a. Aloneness: Extreme in degree and evident soon after birth. The babies do not respond with normal anticipatory gestures when adults reach to pick them up and do not adapt to the bodies of those who hold them.

3. Impaired Communication:

   a. Speech and language are not used for the purposes of communication. Often the children are entirely mute or, if speech is present, it is echolalic and does not convey meaning. Pronominal (relating to a pronoun, in identifying or specifying without describing) reversals and literalness are frequent and affirmation is expressed by repetition rather than the use of the word “yes”.

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4. Obsessive insistence on the maintenance of sameness:
   a. Exhibits and experiences anxiety in new and unfamiliar situations, and engages in ritualistic preoccupation.

5. Fascination for objects, in contrast to disinterest in people:
   a. In some cases, if the early autistic trends are diagnosed and treated early enough, reversals may occur. The prognosis for many autistic children is not very encouraging.

IX. Environmental Influences

A. Sensory Deprivation:

1. To be included under this category requires evidence of atypical parental-child interactions (such as maternal deprivation) or severe environmental restrictions (such as prolonged isolation) during the developmental years.
   a. Infants may display the following behaviors:
      (1) They may become negativistic; refusing to suck and displaying muscular rigidity, shallow breathing, and constipation, or else they regress and become apathetic and depressive, showing no interest in food. An extreme form of regression is marasmus, where the infant becomes lethargic, flabby, and literally wastes away.

2. Psycho-Social Disadvantages:
   a. A criterion for inclusion under this category requires that there be evidence of subnormal intellectual functioning in at least one of the parents and in one or more siblings. These cases are usually from impoverished environments involving poor housing, inadequate diets, and inadequate medical care. There may be prematurity, low-birth weight, or history of infectious diseases, but no single entity appears to have contributed to the slow or retarded development.

X. Other Conditions

This category includes those situations where mental retardation is caused by defects in one or more of the special senses or where there appears to be multiple biological and social conditions contributing to slow or retarded development.
A. Deficit of Special Senses

1. Condition must be the only contributing factor to retardation, example:
   a. Blindness
   b. Deafness
   c. Combination of both
Cerebral palsy has been defined as a group of medical conditions, not a disease, characterized by nerve and dysfunctions. It is a nonprogressive disorder of movement or posture beginning in childhood due to a malfunction or damage of the brain. Sometimes cerebral palsy shows itself only by a slight awkward gait, but more often there are serious handicaps such as seizures, the inability to see, hear, speak or learn as other people do, or psychological or behavioral problems. It involves partial paralysis and lack of muscle coordination resulting from a defect, injury or disease of the nerve tissue contained within the skull. Cerebral palsy is not synonymous with mental retardation, but is found in this classified population.

The physiological classification of this movement disorder was determined by the American Academy for Cerebral Palsy in 1956. There are six groupings of cerebral palsy: spasticity, athetosis, rigidity, ataxia, tremor, and mixed.

**TYPES**

A. **SPASTIC**

   The limb muscle is tight, and with sudden attempted movement or stretching, the muscle contracts strongly. For example: tapping the heel cord results in a quick downward movement of the foot. In spastic paralysis, this movement is exaggerated even to the extent that the muscle will continue to contract repetitively. Children who have spasticity have increased deep tendon reflexes of the involved limbs. As the child grows, the spastic muscle becomes shorter (contracture) with resultant deformities of the limbs, pelvis, and spine.

B. **ATTHETOID**

   Involuntary, purposeless movements. Purposeful movements are contorted.

C. **RIGIDITY**

   Rigidity appears to be a severe form of spasticity. So severe, that if you attempt to move the rigid limb, it gives way as if it were a lead pipe or a cogwheel. These people are usually quadriplegic.

D. **ATAXIA**

   Here, there is a lack of balance sensation. A lack of position sense in space, and uncoordinated movement. When walking, the feet are apart, and weaving of the trunk with arms akimbo (hand on hip with elbow turned outward) to balance the body. Ataxic people fall frequently.

E. **TREMOR**

   Tremor is shakiness of the limb involved. Tremor might be noticed in the attempt to use the limb. Continuous tremor at rest is not uncommon in children, as it is in adults with Parkinson’s disease of the brain.
F. MIXED
Usually these people are quadriplegic and have spasticity and athetosis.

ETIOLOGIES OF CEREBRAL PALSY

A. PRENATAL CAUSES
   1. Inherited: Rare; familial spastic paraplegia
   2. Infections in the mother during pregnancy
   3. Lack of oxygen to the fetal brain
   4. Rh incompatibility
   5. Prematurity: This accounts for 33-60 percent of all cases of cerebral palsy in the United States
   6. Metabolic disorders
   7. Unknown causes: 30% of all prenatal cerebral palsy is in the unknown category

B. PERINATAL CAUSES
   1. Birth injury (trauma)
   2. Lack of oxygen

C. POSTNATAL CAUSES
   1. Head injuries: Skull fractures, brain lacerations or hemorrhages, account for 18% of the total of postnatal causes
   2. Brain infections and toxic conditions: 57% of postnatal causes
   3. Brain hemorrhages or clots
   4. Lack of oxygen to the brain (cerebral anoxia)
   5. Brain tumor

ASSOCIATED HANDICAPS

Cerebral palsy can and often does, affect other systems controlled by the brain. These associated handicaps can be more disabling than the motor disorder.

A. ORAL-DENTAL
   1. Difficulty in swallowing
      a. If greater with fluids, spasm of the pharyngeal musculature is suspected
      b. If greater with solids, paralysis of the pharyngeal muscles might be present
   2. Drooling
   3. Teeth grinding
   4. Tooth enamel malformation
   5. Dental caries are more common
B. SPEECH
Some speech or language defect is present in 48 – 49% of people with cerebral palsy. The speech disorders might be due to paralysis if the speech musculature. Affected children may be unable to organize and select speech.

C. HEARING
Hearing loss is most common in athetosis. Only 2% of the spastic people have hearing problems.

D. VISUAL
Many people are farsighted

E. SENSORY DEFECTS
Loss of shape and texture sensation of the hand may occur, especially in spastic hemiplegia.

F. CONVULSIVE DISORDERS
This occurs in 86% of spastic clients and 12% of athetoids. In postnatal cerebral palsy, seizures occur in 55% of the spastic hemiplegics.

G. MENTAL RETARDATION
Approximately 74% of the people with cerebral palsy have mental retardation of some degree. Serious mental retardation is present in at least 50% of the cases. It is important to remember that 25% of the persons experiencing cerebral palsy have normal or above-normal intellects.

H. PERCEPTUAL AND VISUAL-MOTOR DISORDERS
Cerebral palsy clients do not necessarily see things in a distorted manner, but they may see them in an immature way – a developmental lag.

DIAGNOSIS
The diagnosis of cerebral palsy is practically all clinical. It is based upon the history of the pregnancy, the birth any early development of the client. The physician’s examination is also used. Tests in cerebral palsy exclude the possible progressive neurological illnesses or tell us what the brain damage and its extent might be. The following tests are used selectively.

A. Skull x-rays

B. Electroencephalograms
1. Used in the diagnosis of seizure disorders, or to find focal lesions, such as brain tumors or old scar from prior injury
C. Pneumencephalogram
   1. Injections of air into the ventricles of the brain. A displaced ventricle may mean a tumor in the brain

D. Brain scan
   1. Radioactive isotopes are injected into the blood stream and can help localize brain tumors

E. Cerebral arteriograms
   1. A dye is injected into the blood. The x-ray of the skull will then show the blood vessels and pick-up abnormalities of the arteries in the brain.

F. Blood and urine examinations
   1. May pick up chemical abnormalities that might explain the cerebral palsy

TREATMENT

Treatment varies according to the nature and extent of the brain damage. Muscle relaxants may help reduce spasms. Anticonvulsant drugs are necessary when seizures are among the symptoms of the disorder. Orthopedic surgery, casts, braces, and traction can be used to correct some types of disability associated with cerebral palsy. Early muscle training and special exercises often help the client lead a useful, productive life. In summation: use physical therapy, occupational therapy, bracing, orthopedic surgery, and drugs.
Epilepsy, like many other medical terms, comes from the Greek and means “seizures.” It means nothing more than the tendency to have seizures. Seizures have been called spells, attacks, fits, and convulsions. Epilepsy, by itself, does not cause disfigurement or pain, nor does it shorten the life of the individual. The majority of people with epilepsy are normal, healthy persons with average or above-average intelligence. Not all epileptics are retarded, but it is a symptom that frequently accompanies retardation.

Epilepsy is a disorder of the nervous system in which the major symptom is a convulsive seizure, which is the result of a temporary disturbance of the brain impulses. It is sometimes called cerebral dysrhythmia, meaning a disturbance of the brain’s normal rhythm. There are an estimated 1,860,000 persons with epilepsy in the United States, making it the most common organic disorder of the nervous system.

Brain damage or mental deterioration occurs in only a very small percentage of certain types of epilepsy, or in cases that have been grossly neglected over a long period of years.

**TYPES OF SEIZURES**

**A. GRAND MAL**

In major grand mal seizures, the victim usually emits a cry (a result of air being forced rapidly through the larynx), falls to the floor, unconscious. The body movement consists of a tightening of all muscle (tonic movements), followed by relaxing movements of all muscles (clonic movement), and alternating tonic and clonic movements for from several seconds to several minutes. The eyes may roll back and the individual may or may not froth at the mouth. The client may turn pale and cyanotic. Loss of control of certain body functions may occur such as defecation and voiding. The individual is usually very sleepy following the attack, and usually has no memory of the seizure and/or may complain of a headache. These seizures are produced by over activity in widespread areas of the brain that maintain consciousness and regulate movements of the body. The period of seizure activity is referred to as the ictal state, and the subsequent period of relaxation, the post-ictal state.

**B. PETIT MAL**

These seizures are usually seen in children and may disappear as the child reaches adult life. They consist of a sudden, brief loss of contact with the environment lasting a few seconds. After the interruption in consciousness, the child will continue doing whatever he/she was doing just before the attack. These seizures may occur many times each day and are of short duration. During the attack, the child often seems to blink his/her eyes and may slightly jerk his/her arms and legs without falling to the ground. In these seizures, the over activity involves cells over a wide area on both sides of the brain.
C. **FOCAL**

These seizures are produced by over activity of cells in one area of the brain. The seizure affects the functions, which that area controls or regulates.

1. **Psychomotor seizures**  
a. Consists of a performance of repeated complex movements associated with a clouding of consciousness. The attack may last about a minute, and the individual may appear confused. He/she will have no memory of these seizures. They usually, but not always, originate in an area of the brain called the temporal lobe. This area has very complex functions associated with memory, taste, and smell, as well as automatic behavior.

2. **Focal motor seizures**  
a. Consists of sudden jerking movements in one area such as the face, the arm, or the leg. The over activity can spread to adjacent regions of the brain causing the seizure to gradually involve more and more muscles. This is called a “Jacksonian seizure.” Such a seizure may first begin with shaking or jerking of the hand, followed by the forearm, arm, shoulder, face and continuing in this fashion until the entire half of the body is involved. This may lead to a typical grand mal seizure, if the entire brain becomes involved. In this type of seizure, the specific portion of the brain controlling certain muscles may be diseased or irritated.

3. **Focal sensory**  
a. Involves sensations, such as numbness, tingling and coldness. Sometimes, special sensations such as hearing and seeing are involved.

4. **Acquired epilepsy**  
a. Also called symptomatic epilepsy, has a physical cause, such as a brain tumor, injury to the brain at birth or a wound or blow to the head. These injuries irritate the brain and set off abnormal electrical discharge. In a small percentage of cases, this form of epilepsy may be cured by surgery to remove the tumor or repair the injury.

5. **Idiopathic epilepsy**  
a. This is the most common type of epilepsy and usually manifests itself early in life. Eighty percent of victims have their first seizures before the age of eighteen. This type of epilepsy does not seem to be inherited, although there is evidence that a predisposition to it may run in families. The cause is still unknown.
6. Latent epilepsy
   
a. A person who would have seizures, if the precipitating factor were present, but who actually have never had a seizure due to the absence of the precipitant (i.e., a person who never drinks alcohol, but for whom alcohol would be the precipitating factor).

**AURA**

This is a warning that occurs prior to the seizure. Sometimes these warnings are impossible to describe in words. Auras can consist of strange feelings in the abdomen, spots before your eyes, unusual odors or tastes, past memories, anxiety or tenseness. They present at the beginning of the seizure and can often provide a clue to where the brain cell over activity originates. The aura, very often, also serves as a warning to the person to get themselves into a safe position and posture. That is, if one is driving a car and experiences an aura, he/she would park the car and probably lie down in the back seat. About 95% of seizure clients experience some kind of aura, from immediately prior to the attack to 42 and sometimes even 72 hours prior to the seizures.

**DIAGNOSIS**

Arriving at the proper diagnosis of epilepsy depends upon correct interpretation of the many factors gathered form the client’s history, physical examination, and various laboratory procedures. Specific laboratory tests used are: x-rays, blood and urine examinations, and the electroencephalogram. The physical examination will include a special examination of the nervous system. The examination will include testing of muscle strength, the ability to appreciate sensation of hearing, of eyesight, and reflexes, which will give valuable information about the seizures.

**CONDITIONS THAT INCREASE THE FREQUENCY OF SEIZURES**

There are a number of conditions, which can increase the frequency with which seizure activity occurs in an individual with epilepsy. The general watchword for epileptics is moderation in all things. Any deviation from moderation can precipitate seizure activity. Factors, which represent a lack of moderation, include:

A. Irregular use of medication  
B. Types of illnesses, especially those associated with fever  
C. Emotional stress  
D. Restricted activity and idleness  
E. Fatigue and lack of sleep  
F. Menstrual periods  
G. Constipation

The most common cause of increased seizures is irregular use of medication caused by forgetting to take it or by reduction of the dose without medical advice.
TREATMENT

Anticonvulsant drugs are very helpful in the treatment of epilepsy. (See module 25). Although anticonvulsant drugs do not cure epilepsy, they can usually prevent the disabling effects of the disease, so that the patient can hold a responsible position, and lead practically a normal life with few restrictions.

Because of ignorance and superstition, there is still some stigma attached to epilepsy. Health care professionals need to do their part in counteracting this through educational efforts. The client and his family, as well as the general public, should become familiar with all aspects of this disorder. Specific nursing measures to be taken during an epileptic seizure include:

A. Allow the client to have seizure in privacy and without moving
B. Turn client’s head so that fluid will run out of the mouth, naturally
C. Loosen clothing so that client’s movements are unrestricted
D. Do not attempt to insert tongue blade or airway while client engaged in tonic and clonic movements. Wait for post ictal phase, and then turn head and clear airway. If tongue has fallen back and is obstructing airway, pull tongue forward with gauze, insert airway
E. If client has fallen in a hazardous place, i.e., with head underwater, naturally action must be taken to move him to a safe place. If no hazard involved essential not to move client
F. Monitor vitals during post ictal phase
G. During ictal phase, protect limbs from hitting hard objects and causing injury
Genetic counseling is the process, which brings the advances in medical genetics to the persons who can benefit directly from this information. This could be the parent of a child with a birth defect who needs to know if other children they may have would also be affected. Genetic counseling can also be described as a dynamic process involving a discussion of diagnosis, clinical prognosis, risk of occurrence or recurrence of a disorder in a family and the various ways to deal with that risk.

A four-step process used in genetic counseling has been developed by Dr. Allan J. Ebbin, Assistant Director, Genetics Division of the University of Southern California School of Medicine.

A. ESTABLISHING AN ACCURATE DIAGNOSIS

1. The first step in the counseling process is to establish the diagnosis. This is often a difficult and sometimes impossible task. Without a precise diagnosis, the counselor will not be able to give the family specific information on the risk of recurrence. With advances in medicine, specifically in the area of diagnostic techniques, a definitive diagnosis can be made. Cytogenetic and biochemical tests often can pinpoint the specific genetic cause and are a valuable adjunct to other diagnostic procedures.

B. SECURING A DETAILED FAMILY HISTORY

1. A complete and accurate family history is vital to the process. This medical history includes data on those affected by the disorder in question and those not affected. This information is usually obtained from family records and by contacting family members.

C. PROVIDING UP-TO-DATE GENETIC INFORMATION

1. Genetic counselors are now able to offer information and choices in reproductive planning to persons who seek their services. Amniocentesis is one advancement that has made such counseling possible.

D. ESTABLISHING RAPPORT WITH THE CLIENT

1. The genetic counselor must be able to communicate complex information in an understandable way. Language and cultural differences, as well as many different orientations to the meaning of risk, may complicate the situation. Parents may express feelings of grief and guilt that must be dealt with by the counselor. It is a difficult task.
Dr. Ebbin feels that the following groups of people can best benefit from genetic counseling:

1. People who have a birth defect or genetic disorder and are planning a family
2. Couples who have had a child or a relative with a birth defect, mental retardation, or specific genetic disorder
3. Women 35 years or older who are planning a pregnancy
4. Persons who may be carriers of some disorders because they are members of a particular group; example: Jews who may be concerned about Tay-Sach’s disease, and Blacks concerned with Sickle Cell disease.

AMNIOCENTESIS

This is a procedure in which a small amount of amniotic fluid surrounding a fetus is withdrawn by needle through the mother’s abdominal wall. Cells shed by the fetus in the fluid can then be used for certain tests, which can determine specific genetic defects. This procedure is used most often for Down’s syndrome, Erythroblastosis fetalis, and spina bifida. Amniocentesis also can be used to test for at least 60 other genetic disorders, and researchers are constantly striving to increase this number.

Amniocentesis is performed usually about the 16th week after the beginning of the mother’s last menstrual period about four weeks is required for completion of the laboratory tests. Prospective parents should seek this service before the 14th week of pregnancy, so that they can be counseled about the procedure, the limitation of the tests and the risks.

There are minor risks to the mother and fetus. The small risk for miscarriage after the procedure does not seem to be significantly greater than the chance for spontaneous abortion in women over 35 years of age. The risk of fetal injury also appears low and primarily related to the unavoidable needling of the fetus during the procedure. No adverse effects in the child have occurred. An obstetrician performs the actual withdrawal of the fluid. Ultrasonography, using sound waves and thus avoiding the need for x-rays, provides a picture of the position of the fetus and placenta within the uterus prior to withdrawal of the amniotic fluid.

In the long run, the test results from amniocentesis usually provide nine months of peace of mind for prospective parents who know they are at risk of having a child with a genetic defect. Amniocentesis is called the most important application of medical genetics in recent medical history.
BRIEF AUTISM READING LIST

TECHNICAL BACKGROUND


GENERAL BACKGROUND


PARENT NARRATIVES


A. Prenatal Infections

1. Syphilis
   - Cause: Spirochete infection in an untreated mother. Can be detected by serology. There are two types of congenital syphilis: early and late. Early infection occurs before 2 years of age and late infection occurs after 2 years of age.
   - Major diagnostic features are rash, hepatosplenomegaly, anemia, jaundice, and osteochondritis. The child may develop peg shaped permanent teeth and in the late infection develop perforation of the palate.
   - Major diagnostic features are rash, hepatosplenomegaly, anemia, jaundice, and osteochondritis. The child may develop peg shaped permanent teeth and in the late infection develop perforation of the palate.
   - Treatment: Penicillin is the drug of choice.
   - Syphilis is currently on the increase.

2. Rubella
   - Cause: Viral infection during pregnancy. The most severe damage occurs if the infection occurs during the first trimester.
   - Major diagnostic features include congenital heart defects, deafness, cataracts or glaucoma.
   - Mental retardation may not be present or it may range from mild to profound.
   - Treatment: Lens implants can remedy the cataracts. Surgery can repair some of the heart defects.
Slide 4

Cataract

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Slide 5

3. Toxoplasmosis

- Cause: The protozoa, toxoplasma gondii, infects the mother and then infects the fetus. The protozoa is transferred from the mother to the fetus when human contact occurs. Blood tests are very important to stop the spread. Blood tests will show infection or prior infection.

- Major diagnostic features include:
  - Prolonged infection will result in fetal hydrocephalus, chororetinitis, intercranial calcifications, and mental retardation.
  - Acute infection will show hemolytic anemia, jaundice, purpuric rash, and hepatosplenomegaly.
  - Retardation ranges from mild to severe.

- Treatment: Medications that will kill the protozoa, such as pyrimethamine and sulfadiazine.

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Slide 6

B. Prenatal Intoxications

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1. Fetal Alcohol Syndrome

- Alcohol consumption by the pregnant woman exposed the fetus to great risk. A pattern of craniofacial, limb, and cardiovascular defects associated with prenatal onset, growth deficiency, and developmental delay is known as fetal alcohol syndrome.
- The signs of fetal alcohol syndrome can range from slight retardation, usually detected in school later in life, to profound retardation.
- Drooping eyelids and thin upper lips are characteristic of FAS.

2. Kernicterus

- yellow skin; high bilirubin (blood) level
- severe jaundice; treated with blue light
1. Galactosemia

- Inability to metabolize galactose found in milk

2. Tay-Sachs Disease

- Cause: Autosomal recessive gene
- Major Diagnostic Findings: This is a lethal gene disorder. The lipid accumulation occurs months before the progression begins. The child will die around 3 years of age.
- Treatment: There is no treatment available.

3. Hurler’s Syndrome

- Cause: Autosomal recessive gene
- Major Diagnostic Findings: This is a disorder of the connective tissue. There is an accumulation of mucopolysaccharides, evident by the first year of life. Because of the coarse features and skeletal abnormalities, the syndrome is also known as gargoylism.
- Treatment: There is no treatment available.
4. Phenylketonuria

- **Cause:** Autosomal recessive gene.
- **Major Diagnostic Findings:** This is a disorder in which the body can not metabolize the amino acid phenylalanine and forms the toxic chemical phenylpyruvate. There is a musty odor to the urine after milk is ingested. 20% have seizures. There is tremors and atypical behavior. They are hyperactive, have apraxia, poor coordination, and visual and auditory hallucinations. Their skin tone is light and their eyes are blue. If left untreated the person will become severely or profoundly retarded.
- **Treatment:** Phenylalanine restricted diet.

Most infants are tested for PKU at birth. If placed on a phenylalanine free diet, the infant will develop normally. As an adult, phenylalanine can be included in the diet in measured amounts.

5. Hypothyroidism

- **Cause:** A deficiency in human thyroid hormone.
- **Major Diagnostic Findings:** Hypothyroidism can result from congenital absence of the thyroid gland or from an absence or defect in attitude function. The infant may appear normal during the first six months before a gradual decline occurs. The symptoms include delayed growth, hypotonia, and to end in height. Formerly called cretinism.
- **Treatment:** Thyroid hormone replacement.
- **The person on the right is 28 years old.**

D. Gross Brain Disease
Slide 16

1. Sturge-Weber Disease

- Cause: Possible autosomal dominant genetic disorder
- Major Diagnostic Findings: Venous angioma (port wine stain) on the forehead and cheek as well as the buccal mucosa. Calcification within the brain causes neurological damage and seizures. Retardation varies from normal intelligence to severe mental retardation.
- Treatment: Part of the brain is surgically removed in rare cases to control seizures.

Slide 17

2. Tuberous Sclerosis

- Cause: In some instances this is an autosomal dominant transmission whereas in others cases it has a recessive transmission.
- Major Diagnostic Findings: There is a pink to red butterfly pattern of nodules extending from cheek to cheek across the bridge of the nose. There are areas of depigmentation in infants. The person may have lesions of nails, and tumors of the kidney and eye. Tumors occur in the brain.
- Adenoma sebaceum: Facial gland tumors
- Treatment: Control of seizures is the most important part of treatment.

Slide 18

3. von Recklinghausen’s Disease

- Cause: Autosomal dominant genetic disorder
- Major Diagnostic Findings: Patches of increased pigmentation (cafe-au-lait spots) often present at birth. Skin tumors appear in late childhood or early adolescence. Neurofibromas occur in the back in frequent. Mental retardation is not always present in all cases. Neurofibromas tumors along the nerve endings can cause partial or complete loss of the sense when the tumors are disfiguring or cause mechanical damage.
- Elephant man (movie)
E. Unknown Prenatal Influences

1. Microcephaly
   - Cause: Some are autosomal recessive. Many are as a result of prenatal infection or unknown prenatal influences.
   - Major Diagnostic Findings: Head circumference less than 3 standard deviations below expected average during childhood and less than 45 centimeters at maturity. Mental retardation ranges from mild to profound.
   - Treatment: None to change the brain size or cranium. Special schooling to help habilitate the person.

2. Hydrocephaly
   - Cause: Usually no genetic mechanism. Infection has been linked to producing blockages in the circulation of the cerebral spinal fluid.
   - Major Diagnostic Findings: Increased head size. Protruding eyes. Poor motor development. Neurological abnormalities with mental retardation ranging from none to profound. The flow of cerebral spinal fluid is blocked as it circulates in the brain. If the blockage occurs before the bones of the skull have ossified (hardened), then the resulting pressure will expand the size of the cranium. If the blockage occurs after the ossification of the cranium, the brain material is compressed with resulting damage.
   - Treatment: A shunt (drainage tube) can be surgically placed to drain the fluid from the ventricles of the brain to the heart chamber, peritoneum, or kidneys.
3. Myelomeningocele
    severe forms of spina bifida

- **MENINGOCELE**
  - Cause: A herniation of the meningeal sac through a bony defect in the vertebrae. The meningeal sac protrudes through the herniation to form a "bump" under the skin. The "bump" contains cerebral spinal fluid.

- **MYELOMENINGOCELE**
  - Cause: A herniation of the meningeal sac through a bony defect in the vertebrae.
  - The meningeal sac not only contains the cerebral spinal fluid as in a meningocele, but also contains the spinal nerves. The sac protrudes through the herniation and may be the size of a pea or as large as a soccer ball. Paralysis results from where the nerve protrudes.

Spina bifida

- Neural tube defect 1 per 1000 births
- Incomplete development of the brain, spinal chord, and/or protective coverings for these organs

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Slide 22

Slide 23

Slide 24
4. PRADER WILLI

- Unknown Etiology
- Usually small for term babies
- Involvement with chromosome 15
- Obesity onset from infancy to six years
- Retardation severe to mild, usually moderate
- Hypothyroidism
- Diabetes must be assessed
- Sleep Apnea is common
- Underdeveloped genitalia
- Hypotonia

F. Chromosomal Abnormalities

1. Cri-du-Chat Syndrome

- Cause: Deletion of part of the chromosomal material in one of the short arms of the 5q group
- Chromosomal abnormality
- Major Diagnostic Findings: Small head and severe retardation. Retarded facial expression, hypotonia of the neck, micrognathia, and a cat-like cry.
- Treatment: There is no treatment for the syndrome. Specific treatment for various conditions may be done.
Slide 28

2. Trisomy 21

- **Case:** A third or extra number 21 chromosome - Down’s Syndrome

- **Major Diagnostic Findings:**
  - Tall forehead with flattened occipital area; broad short nose; eyes have an epicanthus fold; low set ears; broad short neck; small head size with long relatively thin fingers. 
  - Respiratory problems are common. 
  - Retardation is present in about 70% of the cases. Moderate to profound levels of retardation is the usual outcome.
  - May develop early Alzheimer’s Disease

Slide 29

Fragile X Syndrome

- There is a fragile site on the long arm of the X chromosome.
- Prominent jaw, thickening nasal bridge, large ears, and pale blue irides.
- Hyperactivity
- Severe to Moderate retardation

Same child at 3 and at 22

Slide 30

amniocentesis
Klinefelter’s Syndrome

- **Cause:** Nondisjunction of the X chromosome resulting in multiple X chromosomes in the male person.
- **Major Diagnostic Findings:** Mental retardation is usually mild. The person is usually tall with decreased secondary sexual characteristics. The male person will not be able to reproduce. Personality or character disorders are common.
- **Treatment:** Male hormone treatment.

Turner’s Syndrome

- **Cause:** An absence of one of the X chromosomes in the female person.
- **Major Diagnostic Findings:** Mental retardation is not always present. The person is shorter in stature and has webbing and/or a shortening of the neck. Ears are low set and the eyes have epicanthic folds. There is an absence of secondary sexual characteristics. The female person will not be able to reproduce.
- **Treatment:** Female hormone treatment.
Turner’s Syndrome

Note the webbing on the neck

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Trisomy 13

- Cause: Extra chromosome 13
- Incidence is 1 per 5000 births
- 18% survive the first year. Cardiac defects are common, cleft lip, large fontanel, low set ears.
- Apnea during infancy
- Polycystic kidneys in 31%
- Seizures
- No Treatment

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Trisomy 13

same child at 12
Slide 37

Etiologies that may not have Developmental Disabilities

Slide 38

APERT’S SYNDROME

- Autosomal dominant
- Mental deficiency may be present
- High forehead, shallow orbits, hypertelorism, midface hypoplasia
- Fingers may be short. Syndactyly
- Recurrence risk is low to negligible
- Surgical attempts to normalize cranial bones may be attempted

Slide 39

Progeria Syndrome

- Etiology unknown
- Often occurs from one to two years
- Premature aging
- Fatigue is a major factor
- Heart attack is the usual cause of death due to relentless arterial atheromatosis
- Intellect is not impaired
Progeria

Life span from 1.5 to 17 years of age

Hallermann-Streiff Syndrome

- Gene mutation is the likely cause.
- Respiratory problems are common.
- Characteristic head and face deformities.
- Intellect is not impaired.

Teratogenic Effects
Slide 43

ANAENCEPHALY

- There is defect in the first 28 days where the neural groove fails to close
- Counseling the parents about the low risk of recurrence
- 90% of infants with anaencephaly do not come to term
- All die
- Folic acid will help to prevent neural tube defects.

Slide 44

Cyclopia

- Failure of the face to complete development
- Large proboscis seen above the orbit area
- Usually associated with other defects and will not survive.

Slide 45

Cloverleaf Skull

- Characteristic skull formation. If the child lives the skull retains the cloverleaf shape.
- Intellect may be impaired.
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STOP!!!

Any Questions???
Etiologies Part 2

- Environmental factors
- Infantile autism
- Cerebral palsy
- Epilepsy

Environmental influences

- 12% due to nonbiological causes
- Battered child syndrome

Infantile Autism: diagnostic features

- Aloneness
- Impaired communication
- Ritualistic preoccupation
- Fascination for objects, disinterest in people
- Poor prognosis

http://www.youtube.com/watch?v=f15JexiQt4U
Slide 4

Cerebral Palsy
Nonprogressive medical condition not a disease
- Oral, dental, and speech problems common
- Spastic
- Athetoid
- Rigidity
- Ataxia
- Tremor
- Mixed

Slide 5

CAUSES
- Inherited
- Infection of the mother
- Physical trauma (birth injury)
- Anoxia
- Unknown

Slide 6

Epilepsy
- A symptom often accompanying retardation
- Grand mal
- Petit mal
- Psychomotor
- Focal motor
- Focal sensory
Seizures continued

- Ictal stage
- Aura
- Status epilepticus
- Conditions which may increase frequency
  - Irregular use of meds
  - Fever, illness
  - Emotional stress
  - Fatigue
  - Constipation